

### LET'S TALK GENETICS Screening and Testing for SLO

Presented by Karen Vikstrom, MS, LCGC Genetic Counselor

### **DHCR7 Carrier Frequencies**

 2017 study of 262,399 people No family history Purpose was routine carrier screening

# QUICK FACTS

#### Most common in individuals with Northern European ancestry,

## **1in 40**

CARRIER RATE

n Northern European ancestry, but seen in all ethnic groups

### **DHCR7 Carrier Frequencies**

### AFRICAN AMERICANS

1 in 183

### ASHKENAZI JEWISH

1 in 43

### HISPANIC

1 in 167

### SOUTHERN EUROPEAN

1 in 66

### NORTHERN EUROPEANS

1 in 54

### MIXED/ OTHER CAUCASIAN

1 in 55

### DHCR7 gene codes for enzymye



### 7-DHC reductase

ENZYME MAKES CHOLESTEROL

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	Recessive inheritance = 1 in 4 chance					
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### What is a genetic variant?

### c.964-1G>C c.452G>A c.278C>T

IVS 8 - 1G > CW15 1\* T93M





Testing for

HOW IT WORKS

## SLO syndrome



#### AMNIOCENTESIS

#### QUAD SC REEN

#### **ULTRASOUND**

### Standard Prenatal Testing and Screening Options

### AMNIOCENTESIS

- Diagnostic
- Sterol profile /gene
- Performed 10-12
  weeks in pregnancy

#### QUAD SC REEN

### ULTRASOUND

### Standard Prenatal Testing and Screening Options

- Diagnostic
- Sterol profile/gene
- Performed 1012 weeks in pregnancy

### AMNIOCENTESIS

- Diagnostic
- Sterol profile/gene
- After 14 weeks

#### QUAD SC REEN

### ULTRASOUND

### Standard Prenatal Testing and Screening Options

- Diagnostic
- Sterol profile/gene
- Performed 1012
  weeks in pregnancy

### AMNIOCENTESIS

- Diagnostic
- Sterol profile/gene
- After 14 weeks

### QUAD SC REEN

- Maternal serum at 16 weeks in pregnancy
- Measures markers
- Requires follow -up

### ULTRASOUND

### Standard Prenatal Testing and Screening Options

- Diagnostic
- Sterol profile/gene
- Performed 1012 weeks in pregnancy

### AMNIOCENTESIS

- Diagnostic
- Sterol profile/gene
- After 14 weeks

### QUAD SC REEN

- Maternal serum at 16 weeks in pregnancy
- Measures markers
- Requires follow-up

### ULTRASOUND

- Not diagnostic
- Not specific
- Requires follow -up

### Standard Prenatal Testing and Screening Options

### MATERNAL URINE

### PREIMPLANTATION GENETIC TESTING (PGT)

- Diagnostic (validated) and non-invasive
- 1422 weeks gestation
- Currently available in the UK
- Sample logistics

Institute of Metabolism and Systems Research (IMSR) in Birmingham, England

### Newer Tests

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### PREIMPLANTATION GENETIC TESTING (PGT)

- Diagnos tic\*
- Cost
- Requires IVF
- Specialized Centers

www.sharinghealthygenes.com

### Newer Tests

### **Carrier Screening**

### **AVAILABLE NOW**

- Hundreds of hereditary conditions, \$250
- Do *before* pregnancy
- Expanded gene panel includes SLO gene
- Not all tests are equal



